



ALDOB gene

aldolase, fructose-bisphosphate B

Normal Function

The *ALDOB* gene provides instructions for making the aldolase B enzyme. This enzyme is one of a group of three aldolase enzymes that are responsible for breaking down certain molecules in cells throughout the body. Four identical aldolase B enzymes need to be attached (bound) to each other in a four-enzyme unit called a tetramer to work.

Aldolase B is found primarily in the liver, but it is also present at lower levels in kidney and intestinal cells. Aldolase B is involved in the breakdown (metabolism) of the simple sugar fructose, which is found mostly in fruits and is used in the body for energy. Aldolase B is responsible for the second step in the metabolism of fructose, which breaks down the molecule fructose-1-phosphate into glyceraldehyde and dihydroxyacetone phosphate. To a lesser degree, aldolase B is also involved in the breakdown of the simple sugar glucose.

Health Conditions Related to Genetic Changes

hereditary fructose intolerance

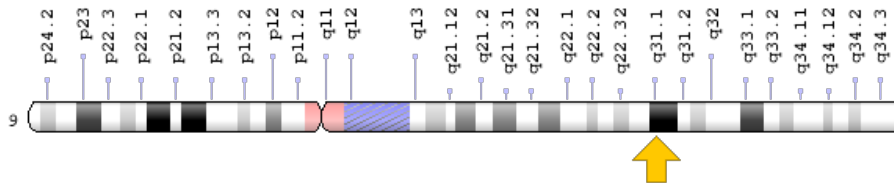
More than 50 mutations in the *ALDOB* gene have been found to cause hereditary fructose intolerance, a condition characterized by nausea and intestinal discomfort following ingestion of foods containing fructose. Most of these mutations replace single protein building blocks (amino acids) in the aldolase B enzyme and result in the production of an enzyme with reduced function. A mutation found in approximately half of people with hereditary fructose intolerance replaces the amino acid alanine with the amino acid proline at position 149 in the enzyme (written as Ala149Pro or A149P). This mutation alters the 3-dimensional shape of the enzyme. Alteration of the shape of the enzyme makes it difficult for the aldolase B enzymes to bind together and form tetramers. If it is not in a tetramer, aldolase B cannot metabolize fructose.

A lack of functional aldolase B results in an accumulation of fructose-1-phosphate in liver cells. This buildup is toxic, resulting in the death of liver cells over time. Additionally, the breakdown products of fructose-1-phosphate are needed in the body to produce energy and to maintain blood sugar levels. The combination of decreased cellular energy, low blood sugar, and liver cell death leads to the features of hereditary fructose intolerance.

Chromosomal Location

Cytogenetic Location: 9q31.1, which is the long (q) arm of chromosome 9 at position 31.1

Molecular Location: base pairs 101,420,560 to 101,435,780 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ALDB
- ALDO2
- ALDOB_HUMAN
- aldolase 2
- aldolase B, fructose-bisphosphatase
- aldolase B, fructose-bisphosphate
- fructose-bisphosphate aldolase B
- liver-type aldolase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Entry of Fructose and Galactose into Glycolysis
<https://www.ncbi.nlm.nih.gov/books/NBK22593/#A2239>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ALDOB%5BTIAB%5D%29+OR+%28aldolase+B%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>

OMIM

- ALDOLASE B, FRUCTOSE-BISPHOSPHATE
<http://omim.org/entry/612724>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/ALDOBID44287ch9q31.html>
- Boston University: Hereditary Fructose Intolerance Mutational Database
<http://www.bu.edu/aldolase/HFI/hfidb/hfidb.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ALDOB%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=417
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/229>
- UniProt
<http://www.uniprot.org/uniprot/P05062>

Sources for This Summary

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